## **Opis choroby \***

## Definicja

A rare multiple congenital anomalies syndrome characterized by cutaneous mastocytosis, microcephaly, microtia and/or hearing loss, hypotonia and skeletal anomalies (e.g. clinodactyly, camptodactyly, scoliosis). Additional common features are short stature, intellectual disability and difficulties. Facial dysmorphism may include upslanted palpebral fissures, highly arched palate and micrognathia. Rarely, seizures and asymmetrically small feet have been reported.

## Dane

Klasyfikacja Zespół wad wrodzonycł	Synonimy Mastocytosis-short stature-deafness syndrome Mastocytoza - niski wzrost - utrata słuchu Mastocytosis-short stature-hearing loss syndrome	
Kod ORPHA 2135	<b>Kod OMIM</b> 248910	<b>Kod ICD10</b> Q82.2
Kod ICD11		

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## <u>\*Źródło</u>

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